

# PAX3

## References

### Sort

- Apuzzo, S. and Gros, P. (2007). Cooperative interactions between the two DNA binding domains of Pax3: helix 2 of the paired domain is in the proximity of the amino terminus of the homeodomain. *Biochemistry*, 46, 2984-93. [\[PubMed\]](#)
- Apuzzo, S., Abdelhakim, A., Fortin, A. S. and Gros, P. (2004). Cross-talk between the paired domain and the homeodomain of Pax3: DNA binding by each domain causes a structural change in the other domain, supporting interdependence for DNA Binding. *J Biol Chem*, 279, 33601-12. [\[PubMed\]](#)
- Apuzzo, S. and Gros, P. (2002). Cooperative interactions between the two DNA binding domains of Pax3: helix 2 of the paired domain is in the proximity of the amino terminus of the homeodomain. *Biochemistry*, 46, 2984-93. [\[PubMed\]](#)
- Asher, J. H., Jr., Harrison, R. W., Morell, R., Carey, M. L. and Friedman, T. B. (1996a). Effects of Pax3 modifier genes on craniofacial morphology, pigmentation, and viability: a murine model of Waardenburg syndrome variation. *Genomics*, 34, 285-98. [\[PubMed\]](#)
- Asher, J. H., Jr., Sommer, A., Morell, R. and Friedman, T. B. (1996b). Missense mutation in the paired domain of PAX3 causes craniofacial-deafness-hand syndrome. *Hum Mutat*, 7, 30-5. [\[PubMed\]](#)
- Auerbach, R. (1954). Analysis of the developmental effects of a lethal mutation in the house mouse. *J. Exp. Zool.*, 127, 305-329.
- Ayme, S. and Philip, N. (1995). Possible homozygous Waardenburg syndrome in a fetus with exencephaly. *Am J Med Genet*, 59, 263-5. [\[PubMed\]](#)
- Bajard, L., Relaix, F., Lagha, M., Rocancourt, D., Daubas, P. and Buckingham, M. E. (2006). A novel genetic hierarchy functions during hypaxial myogenesis: Pax3 directly activates Myf5 in muscle progenitor cells in the limb. *Genes Dev*, 20, 2450-64. [\[PubMed\]](#)
- Baldwin, C. T., Hoth, C. F., Amos, J. A., Da-Silva, E. O. and Milunsky, A. (1992). An exonic mutation in the HuP2 paired domain gene causes Waardenburg's syndrome. *Nature*, 355, 637-8. [\[PubMed\]](#)
- Balling, R., Deutsch, U. and Gruss, P. (1988). Undulated, a mutation affecting the development of the mouse skeleton, has a point mutation in the paired box of Pax 1. *Cell*, 55, 531-5. [\[PubMed\]](#)
- Barber, T. D., Barber, M. C., Cloutier, T. E. and Friedman, T. B. (1999). PAX3 gene structure, alternative splicing and evolution. *Gene*, 237, 311-9. [\[PubMed\]](#)
- Barber, T. D., Barber, M. C., Tomescu, O., Barr, F. G., Ruben, S. and Friedman, T. B. (2002). Identification of target genes regulated by PAX3 and PAX3-FKHR in embryogenesis and alveolar rhabdomyosarcoma. *Genomics*, 79, 278-84. [\[PubMed\]](#)
- Barr, F. G., Fitzgerald, J. C., Ginsberg, J. P., Vanella, M. L., Davis, R. J. and Bennicelli, J. L. (1999). Predominant expression of alternative PAX3 and PAX7 forms in myogenic and neural tumor cell lines. *Cancer Res*, 59, 5443-8. [\[PubMed\]](#)

Barr, F. G., Galili, N., Holick, J., Biegel, J. A., Rovera, G. and Emanuel, B. S. (1993). Rearrangement of the PAX3 paired box gene in the paediatric solid tumour alveolar rhabdomyosarcoma. *Nat Genet*, 3, 113-7. [\[PubMed\]](#)

Beechey, C. V. and Searle, A. G. (1986). Mutations at the Sp locus. *Mouse News Lett* 75, 28.

Betters, E., Liu, Y., Kjaeldgaard, A., Sundstrom, E. and Garcia-Castro, M. (2010). Analysis of early human neural crest development. *Dev Biol*. [\[PubMed\]](#)

Birrane, G., Soni, A. and Ladias, J. A. (2009). Structural Basis for DNA Recognition by the Human PAX3 Homeodomain (dagger) (,) (double dagger). *Biochemistry*. [\[PubMed\]](#)

Blake, J. A. and Ziman, M. R. (2005). Pax3 transcripts in melanoblast development. *Dev Growth Differ*, 47, 627-35. [\[PubMed\]](#)

Bogani, D., Warr, N., Elms, P., Davies, J., Tymowska-Lalanne, Z., Goldsworthy, M., Cox, R. D., Keays, D. A., Flint, J., Wilson, V., et al. (2004). New semidominant mutations that affect mouse development. *Genesis*, 40, 109-117. [\[PubMed\]](#)

Bondurand, N., Pingault, V., Goerich, D. E., Lemort, N., Sock, E., Le Caignec, C., Wegner, M. and Goossens, M. (2000). Interaction among SOX10, PAX3 and MITF, three genes altered in Waardenburg syndrome. *Hum Mol Genet*, 9, 1907-17. [\[PubMed\]](#)

Brown, C. B., Engleka, K. A., Wenning, J., Min Lu, M. and Epstein, J. A. (2005). Identification of a hypaxial somite enhancer element regulating Pax3 expression in migrating myoblasts and characterization of hypaxial muscle Cre transgenic mice. *Genesis*, 41, 202-9. [\[PubMed\]](#)

Burri, M., Tromvoukis, Y., Bopp, D., Frigerio, G. and Noll, M. (1989). Conservation of the paired domain in metazoans and its structure in three isolated human genes. *EMBO J*, 8, 1183-90. [\[PubMed\]](#)

Cao, Y. and Wang, C. (2000). The COOH-terminal transactivation domain plays a key role in regulating the in vitro and in vivo function of Pax3 homeodomain. *J Biol Chem*, 275, 9854-62. [\[PubMed\]](#)

Cattanach, B. M., Beechey, C. V., Rasberry, C. and Evans, E. P. (1994). Mutations Pax and Pax. *Mouse Genome*, 92, 503-4.

Chalepakis, G., Goulding, M., Read, A., Strachan, T. and Gruss, P. (1994a). Molecular basis of splotch and Waardenburg Pax-3 mutations. *Proc Natl Acad Sci U S A*, 91, 3685-9. [\[PubMed\]](#)

Chalepakis, G., Jones, F. S., Edelman, G. M. and Gruss, P. (1994b). Pax-3 contains domains for transcription activation and transcription inhibition. *Proc Natl Acad Sci U S A*, 91, 12745-9. [\[PubMed\]](#)

Chang, T. I., Horal, M., Jain, S. K., Wang, F., Patel, R. and Loeken, M. R. (2003). Oxidant regulation of gene expression and neural tube development: Insights gained from diabetic pregnancy on molecular causes of neural tube defects. *Diabetologia*, 46, 538-45. [\[PubMed\]](#)

Chi, N. and Epstein, J. A. (2002). Getting your Pax straight: Pax proteins in development and disease. *Trends Genet*, 18, 41-7. [\[PubMed\]](#)

Corry, G. N., Hendzel, M. J. and Underhill, D. A. (2008). Subnuclear localization and mobility are key indicators of PAX3 dysfunction in Waardenburg syndrome. *Hum Mol Genet*, 17, 1825-37. [\[PubMed\]](#)

Corry, G. N. and Underhill, D. A. (2005). Pax3 target gene recognition occurs through distinct modes that are differentially affected by disease-associated mutations. *Pigment Cell Res*, 18, 427-38. [\[PubMed\]](#)

- Degenhardt, K. R., Milewski, R. C., Padmanabhan, A., Miller, M., Singh, M. K., Lang, D., Engleka, K. A., Wu, M., Li, J., Zhou, D., et al. (2010). Distinct enhancers at the Pax3 locus can function redundantly to regulate neural tube and neural crest expressions. *Developmental biology*, 339, 519-27. [\[PubMed\]](#)
- Dickie, M. M. (1964). New Splotch Alleles in the Mouse. *J Hered*, 55, 97-101. [\[PubMed\]](#)
- Engleka, K. A., Gitler, A. D., Zhang, M., Zhou, D. D., High, F. A. and Epstein, J. A. (2005). Insertion of Cre into the Pax3 locus creates a new allele of Splotch and identifies unexpected Pax3 derivatives. *Dev Biol*, 280, 396-406. [\[PubMed\]](#)
- Epstein, D. J., Malo, D., Vekemans, M. and Gros, P. (1991a). Molecular characterization of a deletion encompassing the splotch mutation on mouse chromosome 1. *Genomics*, 10, 89-93. [\[PubMed\]](#)
- Epstein, D. J., Vekemans, M. and Gros, P. (1991b). Splotch (Sp2H), a mutation affecting development of the mouse neural tube, shows a deletion within the paired homeodomain of Pax-3. *Cell*, 67, 767-74. [\[PubMed\]](#)
- Epstein, J. A., Shapiro, D. N., Cheng, J., Lam, P. Y. and Maas, R. L. (1996). Pax3 modulates expression of the c-Met receptor during limb muscle development. *Proc Natl Acad Sci U S A*, 93, 4213-8. [\[PubMed\]](#)
- Farrer, L. A., Arnos, K. S., Asher, J. H., Jr., Baldwin, C. T., Diehl, S. R., Friedman, T. B., Greenberg, J., Grundfast, K. M., Hoth, C., Lalwani, A. K., et al. (1994). Locus heterogeneity for Waardenburg syndrome is predictive of clinical subtypes. *Am J Hum Genet*, 55, 728-37. [\[PubMed\]](#)
- Fenby, B. T., Fotaki, V. and Mason, J. O. (2008). Pax3 regulates Wnt1 expression via a conserved binding site in the 5' proximal promoter. *Biochim Biophys Acta*, 1779, 115-21. [\[PubMed\]](#)
- Fine, E. L., Horal, M., Chang, T. I., Fortin, G. and Loeken, M. R. (1999). Evidence that elevated glucose causes altered gene expression, apoptosis, and neural tube defects in a mouse model of diabetic pregnancy. *Diabetes*, 48, 2454-62. [\[PubMed\]](#)
- Fortin, A. S., Underhill, D. A. and Gros, P. (1997). Reciprocal effect of Waardenburg syndrome mutations on DNA binding by the Pax-3 paired domain and homeodomain. *Hum Mol Genet*, 6, 1781-90. [\[PubMed\]](#)
- Franz, T. and Kothary, R. (1993). Characterization of the neural crest defect in Splotch (Sp1H) mutant mice using a lacZ transgene. *Brain Res Dev Brain Res*, 72, 99-105. [\[PubMed\]](#)
- Galibert, M. D., Yavuzer, U., Dexter, T. J. and Goding, C. R. (1999). Pax3 and regulation of the melanocyte-specific tyrosinase-related protein-1 promoter. *J Biol Chem*, 274, 26894-900. [\[PubMed\]](#)
- Galili, N., Davis, R. J., Fredericks, W. J., Mukhopadhyay, S., Rauscher, F. J., 3rd, Emanuel, B. S., Rovera, G. and Barr, F. G. (1993). Fusion of a fork head domain gene to PAX3 in the solid tumour alveolar rhabdomyosarcoma. *Nat Genet*, 5, 230-5. [\[PubMed\]](#)
- Gershon, T. R., Oppenheimer, O., Chin, S. S. and Gerald, W. L. (2005). Temporally regulated neural crest transcription factors distinguish neuroectodermal tumors of varying malignancy and differentiation. *Neoplasia*, 7, 575-84. [\[PubMed\]](#)
- Glaser, T., Walton, D. S. and Maas, R. L. (1992). Genomic structure, evolutionary conservation and aniridia mutations in the human PAX6 gene. *Nat Genet*, 2, 232-9. [\[PubMed\]](#)
- Guo, X. L., Ruan, H. B., Li, Y., Gao, X. and Li, W. (2010). Identification of a novel nonsense mutation on the Pax3 gene in ENU-derived white belly spotting mice and its genetic interaction with c-Kit. *Pigment Cell Melanoma Res*, 23, 252-62. [\[PubMed\]](#)

- Goulding, M. D., Chalepakis, G., Deutsch, U., Erselius, J. R. and Gruss, P. (1991). Pax-3, a novel murine DNA binding protein expressed during early neurogenesis. *EMBO J*, 10, 1135-47. [\[PubMed\]](#)
- Goulding, M. D., Lumsden, A. and Gruss, P. (1993). Signals from the notochord and floor plate regulate the region-specific expression of two Pax genes in the developing spinal cord. *Development*, 117, 1001-16. [\[PubMed\]](#)
- He, S. J., Stevens, G., Braithwaite, A. W. and Eccles, M. R. (2005). Transfection of melanoma cells with antisense PAX3 oligonucleotides additively complements cisplatin-induced cytotoxicity. *Mol Cancer Ther*, 4, 996-1003. [\[PubMed\]](#)
- He, S., Yoon, H. S., Suh, B. J. and Eccles, M. R. (2010). PAX3 Is extensively expressed in benign and malignant tissues of the melanocytic lineage in humans. *J Invest Dermatol*, 130, 1465-8. [\[PubMed\]](#)
- Henderson, D. J., Ybot-Gonzalez, P. and Copp, A. J. (1997). Over-expression of the chondroitin sulphate proteoglycan versican is associated with defective neural crest migration in the Pax3 mutant mouse (splotch). *Mech Dev*, 69, 39-51. [\[PubMed\]](#)
- Hilari, J. M., Mangas, C., Xi, L., Paradelo, C., Ferrandiz, C., Hughes, S. J., Yueh, C., Altomare, I., Gooding, W. E. and Godfrey, T. E. (2009). Molecular staging of pathologically negative sentinel lymph nodes from melanoma patients using multimarker, quantitative real-time rt-PCR. *Ann Surg Oncol*, 16, 177-85. [\[PubMed\]](#)
- Hill, A. L., Phelan, S. A. and Loeken, M. R. (1998). Reduced expression of pax-3 is associated with overexpression of cdc46 in the mouse embryo. *Dev Genes Evol*, 208, 128-34. [\[PubMed\]](#)
- Hill, R. E., Favor, J., Hogan, B. L., Ton, C. C., Saunders, G. F., Hanson, I. M., Prosser, J., Jordan, T., Hastie, N. D. and Van Heyningen, V. (1991). Mouse small eye results from mutations in a paired-like homeobox-containing gene. *Nature*, 354, 522-5. [\[PubMed\]](#)
- Hollenbach, A. D., Sublett, J. E., Mcpherson, C. J. and Grosveld, G. (1999). The Pax3-FKHR oncprotein is unresponsive to the Pax3-associated repressor hDaxx. *EMBO J*, 18, 3702-11. [\[PubMed\]](#)
- Hornyak, T. J., Hayes, D. J., Chiu, L. Y. and Ziff, E. B. (2001). Transcription factors in melanocyte development: distinct roles for Pax-3 and Mitf. *Mech Dev*, 101, 47-59. [\[PubMed\]](#)
- Hoth, C. F., Milunsky, A., Lipsky, N., Sheffer, R., Clarren, S. K. and Baldwin, C. T. (1993). Mutations in the paired domain of the human PAX3 gene cause Klein-Waardenburg syndrome (WS-III) as well as Waardenburg syndrome type I (WS-I). *Am J Hum Genet*, 52, 455-62. [\[PubMed\]](#)
- Hou, L., Loftus, S. K., Incao, A., Chen, A. and Pavan, W. J. (2004). Complementation of melanocyte development in SOX10 mutant neural crest using lineage-directed gene transfer. *Dev Dyn*, 229, 54-62. [\[PubMed\]](#)
- Hsieh, M. J., Yao, Y. L., Lai, I. L. and Yang, W. M. (2006). Transcriptional repression activity of PAX3 is modulated by competition between corepressor KAP1 and heterochromatin protein 1. *Biochem Biophys Res Commun*, 349, 573-81. [\[PubMed\]](#)
- Jordan, T., Hanson, I., Zaletayev, D., Hodgson, S., Prosser, J., Seawright, A., Hastie, N. and Van Heyningen, V. (1992). The human PAX6 gene is mutated in two patients with aniridia. *Nat Genet*, 1, 328-32. [\[PubMed\]](#)
- Kamaraju, A. K., Adjalley, S., Zhang, P., Chebath, J. and Revel, M. (2004). C/EBP-delta induction by gp130 signaling. Role in transition to myelin gene expressing phenotype in a melanoma cell line model. *J Biol Chem*, 279, 3852-61. [\[PubMed\]](#)

Kamaraju, A. K., Bertolotto, C., Chebath, J. and Revel, M. (2002). Pax3 down-regulation and shut-off of melanogenesis in melanoma B16/F10.9 by interleukin-6 receptor signaling. *J Biol Chem*, 277, 15132-41. [\[PubMed\]](#)

Keller, C., Arenkiel, B. R., Coffin, C. M., El-Bardeesy, N., Depinho, R. A. and Capecchi, M. R. (2004). Alveolar rhabdomyosarcomas in conditional Pax3:Fkhr mice: cooperativity of Ink4a/ARF and Trp53 loss of function. *Genes Dev*, 18, 2614-26. [\[PubMed\]](#)

Koushik, S. V., Chen, H., Wang, J. and Conway, S. J. (2002). Generation of a conditional loxP allele of the Pax3 transcription factor that enables selective deletion of the homeodomain. *Genesis*, 32, 114-7. [\[PubMed\]](#)

Koyanagi, K., Kuo, C., Nakagawa, T., Mori, T., Ueno, H., Lorigo, A. R., Jr., Wang, H. J., Hseuh, E., O'day, S. J. and Hoon, D. S. (2005a). Multimarker quantitative real-time PCR detection of circulating melanoma cells in peripheral blood: relation to disease stage in melanoma patients. *Clin Chem*, 51, 981-8. [\[PubMed\]](#)

Koyanagi, K., O'day, S. J., Gonzalez, R., Lewis, K., Robinson, W. A., Amatruda, T. T., Wang, H. J., Elashoff, R. M., Takeuchi, H., Umetani, N., et al. (2005b). Serial monitoring of circulating melanoma cells during neoadjuvant biochemotherapy for stage III melanoma: outcome prediction in a multicenter trial. *J Clin Oncol*, 23, 8057-64. [\[PubMed\]](#)

Lacosta, A. M., Canudas, J., Gonzalez, C., Muniesa, P., Sarasa, M. and Dominguez, L. (2007). Pax7 identifies neural crest, chromatophore lineages and pigment stem cells during zebrafish development. *Int J Dev Biol*, 51, 327-31. [\[PubMed\]](#)

Lacosta, A. M., Muniesa, P., Ruberte, J., Sarasa, M. and Dominguez, L. (2005). Novel expression patterns of Pax3/Pax7 in early trunk neural crest and its melanocyte and non-melanocyte lineages in amniote embryos. *Pigment Cell Res*, 18, 243-51. [\[PubMed\]](#)

Lagutina, I., Conway, S. J., Sublett, J. and Grosveld, G. C. (2002). Pax3-FKHR knock-in mice show developmental aberrations but do not develop tumors. *Mol Cell Biol*, 22, 7204-16. [\[PubMed\]](#)

Lai, I. L., Lin, T. P., Yao, Y. L., Lin, C. Y., Hsieh, M. J. and Yang, W. M. (2010). Histone deacetylase 10 relieves repression on the melanogenic program by maintaining the deacetylation status of repressors. *J Biol Chem*, 285, 7187-96. [\[PubMed\]](#)

Lang, D., Chen, F., Milewski, R., Li, J., Lu, M. M. and Epstein, J. A. (2000). Pax3 is required for enteric ganglia formation and functions with Sox10 to modulate expression of c-ret. *J Clin Invest*, 106, 963-71. [\[PubMed\]](#)

Lang, D. and Epstein, J. A. (2003). Sox10 and Pax3 physically interact to mediate activation of a conserved c-RET enhancer. *Hum Mol Genet*, 12, 937-45. [\[PubMed\]](#)

Lang, D., Lu, M. M., Huang, L., Engleka, K. A., Zhang, M., Chu, E. Y., Lipner, S., Skoultchi, A., Millar, S. E. and Epstein, J. A. (2005). Pax3 functions at a nodal point in melanocyte stem cell differentiation. *Nature*, 433, 884-7. [\[PubMed\]](#)

Lee, M., Goodall, J., Verastegui, C., Ballotti, R. and Goding, C. R. (2000). Direct regulation of the Microphthalmia promoter by Sox10 links Waardenburg-Shah syndrome (WS4)-associated hypopigmentation and deafness to WS2. *J Biol Chem*, 275, 37978-83. [\[PubMed\]](#)

Li, H. G., Wang, Q., Li, H. M., Kumar, S., Parker, C., Slevin, M. and Kumar, P. (2007). PAX3 and PAX3-FKHR promote rhabdomyosarcoma cell survival through downregulation of PTEN. *Cancer Lett*, 253, 215-23. [\[PubMed\]](#)

Li, J., Chen, F. and Epstein, J. A. (2000). Neural crest expression of Cre recombinase directed by the proximal Pax3 promoter in transgenic mice. *Genesis*, 26, 162-4. [\[PubMed\]](#)

Li, J., Liu, K. C., Jin, F., Lu, M. M. and Epstein, J. A. (1999). Transgenic rescue of congenital heart disease and spina bifida in Splotch mice. *Development*, 126, 2495-503. [\[PubMed\]](#)

Magnaghi, P., Roberts, C., Lorain, S., Lipinski, M. and Scambler, P. J. (1998). HIRA, a mammalian homologue of *Saccharomyces cerevisiae* transcriptional co-repressors, interacts with Pax3. *Nat Genet*, 20, 74-7. [\[PubMed\]](#)

Mansouri, A., Pla, P., Larue, L. and Gruss, P. (2001). Pax3 acts cell autonomously in the neural tube and somites by controlling cell surface properties. *Development*, 128, 1995-2005. [\[PubMed\]](#)

Margue, C. M., Bernasconi, M., Barr, F. G. and Schafer, B. W. (2000). Transcriptional modulation of the anti-apoptotic protein BCL-XL by the paired box transcription factors PAX3 and PAX3/FKHR. *Oncogene*, 19, 2921-9. [\[PubMed\]](#)

Mascarenhas, J. B., Littlejohn, E. L., Wolsky, R. J., Young, K. P., Nelson, M., Salgia, R. and Lang, D. (2010). PAX3 and SOX10 activate MET receptor expression in melanoma. *Pigment Cell Melanoma Res*, 23, 225-37. [\[PubMed\]](#)

Matsuzaki, Y., Hashimoto, S., Fujita, T., Suzuki, T., Sakurai, T., Matsushima, K. and Kawakami, Y. (2005). Systematic identification of human melanoma antigens using serial analysis of gene expression (SAGE). *J Immunother*, 28, 10-9. [\[PubMed\]](#)

Mayanil, C. S., George, D., Freilich, L., Miljan, E. J., Mania-Farnell, B., Mcclone, D. G. and Bremer, E. G. (2001). Microarray analysis detects novel Pax3 downstream target genes. *J Biol Chem*, 276, 49299-309. [\[PubMed\]](#)

Mayanil, C. S., Pool, A., Nakazaki, H., Reddy, A. C., Mania-Farnell, B., Yun, B., George, D., Mcclone, D. G. and Bremer, E. G. (2006). Regulation of murine TGFbeta2 by Pax3 during early embryonic development. *J Biol Chem*, 281, 24544-52. [\[PubMed\]](#)

Medic, S., Rizos, H. and Ziman, M. (2011). Differential PAX3 functions in normal skin melanocytes and melanoma cells. *Biochemical and biophysical research communications*, 411, 832-7. [\[PubMed\]](#)

Medic, S. and Ziman, M. (2010). PAX3 expression in normal skin melanocytes and melanocytic lesions (naevi and melanomas). *PLoS One*, 5, e9977. [\[PubMed\]](#)

Milewski, R. C., Chi, N. C., Li, J., Brown, C., Lu, M. M. and Epstein, J. A. (2004). Identification of minimal enhancer elements sufficient for Pax3 expression in neural crest and implication of Tead2 as a regulator of Pax3. *Development*, 131, 829-37. [\[PubMed\]](#)

Milunsky, J. M., Maher, T. A., Ito, M. and Milunsky, A. (2007). The value of MLPA in Waardenburg syndrome. *Genet Test*, 11, 179-82. [\[PubMed\]](#)

Minchin, J. E. and Hughes, S. M. (2008). Sequential actions of Pax3 and Pax7 drive xanthophore development in zebrafish neural crest. *Dev Biol*, 317, 508-22. [\[PubMed\]](#)

Murakami, M., Tominaga, J., Makita, R., Uchijima, Y., Kurihara, Y., Nakagawa, O., Asano, T. and Kurihara, H. (2006). Transcriptional activity of Pax3 is co-activated by TAZ. *Biochem Biophys Res Commun*, 339, 533-9. [\[PubMed\]](#)

- Muratovska, A., Zhou, C., He, S., Goodyer, P. and Eccles, M. R. (2003). Paired-Box genes are frequently expressed in cancer and often required for cancer cell survival. *Oncogene*, 22, 7989-97. [\[PubMed\]](#)
- Nakazaki, H., Reddy, A. C., Mania-Farnell, B. L., Shen, Y. W., Ichi, S., McCabe, C., George, D., Mcclone, D. G., Tomita, T. and Mayanil, C. S. (2008). Key basic helix-loop-helix transcription factor genes Hes1 and Ngn2 are regulated by Pax3 during mouse embryonic development. *Dev Biol*, 316, 510-23. [\[PubMed\]](#)
- Nakazaki, H., Shen, Y. W., Yun, B., Reddy, A., Khanna, V., Mania-Farnell, B., Ichi, S., Mcclone, D. G., Tomita, T. and Mayanil, C. S. (2009). Transcriptional regulation by Pax3 and TGFbeta2 signaling: a potential gene regulatory network in neural crest development. *Int J Dev Biol*, 53, 69-79. [\[PubMed\]](#)
- Natoli, T. A., Ellsworth, M. K., Wu, C., Gross, K. W. and Pruitt, S. C. (1997). Positive and negative DNA sequence elements are required to establish the pattern of Pax3 expression. *Development*, 124, 617-26. [\[PubMed\]](#)
- Pani, L., Horal, M. and Loeken, M. R. (2002). Rescue of neural tube defects in Pax-3-deficient embryos by p53 loss of function: implications for Pax-3-dependent development and tumorigenesis. *Genes Dev*, 16, 676-80. [\[PubMed\]](#)
- Parker, C. J., Shawcross, S. G., Li, H., Wang, Q. Y., Herrington, C. S., Kumar, S., Mackie, R. M., Prime, W., Rennie, I. G., Sisley, K., et al. (2004). Expression of PAX 3 alternatively spliced transcripts and identification of two new isoforms in human tumors of neural crest origin. *Int J Cancer*, 108, 314-20. [\[PubMed\]](#)
- Phelan, S. A., Ito, M. and Loeken, M. R. (1997). Neural tube defects in embryos of diabetic mice: role of the Pax-3 gene and apoptosis. *Diabetes*, 46, 1189-97. [\[PubMed\]](#)
- Ploski, J. E., Shamsher, M. K. and Radu, A. (2004). Paired-type homeodomain transcription factors are imported into the nucleus by karyopherin 13. *Mol Cell Biol*, 24, 4824-34. [\[PubMed\]](#)
- Plummer, R. S., Shea, C. R., Nelson, M., Powell, S. K., Freeman, D. M., Dan, C. P. and Lang, D. (2008). PAX3 expression in primary melanomas and nevi. *Mod Pathol*, 21, 525-30. [\[PubMed\]](#)
- Potterf, S. B., Furumura, M., Dunn, K. J., Arnheiter, H. and Pavan, W. J. (2000). Transcription factor hierarchy in Waardenburg syndrome: regulation of MITF expression by SOX10 and PAX3. *Hum Genet*, 107, 1-6. [\[PubMed\]](#)
- Pruitt, S. C., Bussman, A., Maslov, A. Y., Natoli, T. A. and Heinaman, R. (2004). Hox/Pbx and Brn binding sites mediate Pax3 expression in vitro and in vivo. *Gene Expr Patterns*, 4, 671-85. [\[PubMed\]](#)
- Rasberry, C. and Cattanach, B. M. (1994). A new mutation at the Ph locus. *Mouse Genome*, 92, 504-5.
- Read, A. P. and Newton, V. E. (1997). Waardenburg syndrome. *J Med Genet*, 34, 656-65. [\[PubMed\]](#)
- Relaix, F., Polimeni, M., Rocancourt, D., Ponzetto, C., Schafer, B. W. and Buckingham, M. (2003). The transcriptional activator PAX3-FKHR rescues the defects of Pax3 mutant mice but induces a myogenic gain-of-function phenotype with ligand-independent activation of Met signaling in vivo. *Genes Dev*, 17, 2950-65. [\[PubMed\]](#)
- Relaix, F., Rocancourt, D., Mansouri, A. and Buckingham, M. (2004). Divergent functions of murine Pax3 and Pax7 in limb muscle development. *Genes Dev*, 18, 1088-105. [\[PubMed\]](#)
- Relaix, F., Rocancourt, D., Mansouri, A. and Buckingham, M. (2005). A Pax3/Pax7-dependent population of skeletal muscle progenitor cells. *Nature*, 435, 948-53. [\[PubMed\]](#)

- Rodeberg, D. A., Nuss, R. A., Elsawa, S. F., Erskine, C. L. and Celis, E. (2006). Generation of tumoricidal PAX3 peptide antigen specific cytotoxic T lymphocytes. *Int J Cancer*, 119, 126-32. [\[PubMed\]](#)
- Russell, W. L. and Roscoe, B. (1947). Splotch, a new mutation in the house mouse. *Genetics*, 32, 102.
- Ryu, B., Kim, D. S., Deluca, A. M. and Alani, R. M. (2007). Comprehensive expression profiling of tumor cell lines identifies molecular signatures of melanoma progression. *PLoS ONE*, 2, e594. [\[PubMed\]](#)
- Scholl, F. A., Kamarashev, J., Murmann, O. V., Geertsen, R., Dummer, R. and Schafer, B. W. (2001). PAX3 is expressed in human melanomas and contributes to tumor cell survival. *Cancer Res*, 61, 823-6. [\[PubMed\]](#)
- Shapiro, D. N., Sublett, J. E., Li, B., Downing, J. R. and Naeve, C. W. (1993). Fusion of PAX3 to a member of the forkhead family of transcription factors in human alveolar rhabdomyosarcoma. *Cancer Res*, 53, 5108-12. [\[PubMed\]](#)
- Smit, D. J., Smith, A. G., Parsons, P. G., Muscat, G. E. and Sturm, R. A. (2000). Domains of Brn-2 that mediate homodimerization and interaction with general and melanocytic transcription factors. *Eur J Biochem*, 267, 6413-22. [\[PubMed\]](#)
- Sommer, A. and Bartholomew, D. W. (2003). Craniofacial-deafness-hand syndrome revisited. *Am J Med Genet A*, 123A, 91-4. [\[PubMed\]](#)
- Sommer, A., Young-Wee, T. and Frye, T. (1983). Previously undescribed syndrome of craniofacial, hand anomalies, and sensorineural deafness. *Am J Med Genet*, 15, 71-7. [\[PubMed\]](#)
- Stoller, J. Z., Degenhardt, K. R., Huang, L., Zhou, D. D., Lu, M. M. and Epstein, J. A. (2008). Cre reporter mouse expressing a nuclear localized fusion of GFP and beta-galactosidase reveals new derivatives of Pax3-expressing precursors. *Genesis*, 46, 200-4. [\[PubMed\]](#)
- Takeuchi, H., Morton, D. L., Kuo, C., Turner, R. R., Elashoff, D., Elashoff, R., Taback, B., Fujimoto, A. and Hoon, D. S. (2004). Prognostic significance of molecular upstaging of paraffin-embedded sentinel lymph nodes in melanoma patients. *J Clin Oncol*, 22, 2671-80. [\[PubMed\]](#)
- Tassabehji, M., Newton, V. E., Leverton, K., Turnbull, K., Seemanova, E., Kunze, J., Sperling, K., Strachan, T. and Read, A. P. (1994). PAX3 gene structure and mutations: close analogies between Waardenburg syndrome and the Splotch mouse. *Hum Mol Genet*, 3, 1069-74. [\[PubMed\]](#)
- Tassabehji, M., Newton, V. E., Liu, X. Z., Brady, A., Donnai, D., Krajewska-Walasek, M., Murday, V., Norman, A., Oberszty, E., Reardon, W., et al. (1995). The mutational spectrum in Waardenburg syndrome. *Hum Mol Genet*, 4, 2131-7. [\[PubMed\]](#)
- Tassabehji, M., Read, A. P., Newton, V. E., Harris, R., Balling, R., Gruss, P. and Strachan, T. (1992). Waardenburg's syndrome patients have mutations in the human homologue of the Pax-3 paired box gene. *Nature*, 355, 635-6. [\[PubMed\]](#)
- Tassabehji, M., Read, A. P., Newton, V. E., Patton, M., Gruss, P., Harris, R. and Strachan, T. (1993). Mutations in the PAX3 gene causing Waardenburg syndrome type 1 and type 2. *Nat Genet*, 3, 26-30. [\[PubMed\]](#)
- Tatzel, J., Poser, I., Schroeder, J. and Bosserhoff, A. K. (2005). Inhibition of melanoma inhibitory activity (MIA) expression in melanoma cells leads to molecular and phenotypic changes. *Pigment Cell Res*, 18, 92-101. [\[PubMed\]](#)
- Tekin, M., Bodurtha, J. N., Nance, W. E. and Pandya, A. (2001). Waardenburg syndrome type 3

(Klein-Waardenburg syndrome) segregating with a heterozygous deletion in the paired box domain of PAX3: a simple variant or a true syndrome? *Clin Genet*, 60, 301-4. [\[PubMed\]](#)

Thomas, A. J. and Erickson, C. A. (2009). FOXD3 regulates the lineage switch between neural crest-derived glial cells and pigment cells by repressing MITF through a non-canonical mechanism. *Development*, 136, 1849-58. [\[PubMed\]](#)

Torres, M., Gomez-Pardo, E., Dressler, G. R. and Gruss, P. (1995). Pax-2 controls multiple steps of urogenital development. *Development*, 121, 4057-65. [\[PubMed\]](#)

Tshori, S., Gilon, D., Beeri, R., Nechushtan, H., Kaluzhny, D., Pikarsky, E. and Razin, E. (2006). Transcription factor MITF regulates cardiac growth and hypertrophy. *J Clin Invest*, 116, 2673-81. [\[PubMed\]](#)

Tsukamoto, K., Nakamura, Y. and Niikawa, N. (1994). Isolation of two isoforms of the PAX3 gene transcripts and their tissue-specific alternative expression in human adult tissues. *Hum Genet*, 93, 270-4. [\[PubMed\]](#)

Underhill, D. A. and Gros, P. (1997). The paired-domain regulates DNA binding by the homeodomain within the intact Pax-3 protein. *J Biol Chem*, 272, 14175-82. [\[PubMed\]](#)

Underhill, D. A., Vogan, K. J. and Gros, P. (1995). Analysis of the mouse Splotch-delayed mutation indicates that the Pax-3 paired domain can influence homeodomain DNA-binding activity. *Proc Natl Acad Sci U S A*, 92, 3692-6. [\[PubMed\]](#)

Underwood, T. J., Amin, J., Lillycrop, K. A. and Blaydes, J. P. (2007). Dissection of the functional interaction between p53 and the embryonic proto-oncogene PAX3. *FEBS Lett*, 581, 5831-5. [\[PubMed\]](#)

Vachtenheim, J. and Novotna, H. (1999). Expression of genes for microphthalmia isoforms, Pax3 and MSG1, in human melanomas. *Cell Mol Biol (Noisy-le-grand)*, 45, 1075-82. [\[PubMed\]](#)

Verastegui, C., Bille, K., Ortonne, J. P. and Ballotti, R. (2000). Regulation of the microphthalmia-associated transcription factor gene by the Waardenburg syndrome type 4 gene, SOX10. *J Biol Chem*, 275, 30757-60. [\[PubMed\]](#)

Wang, Q., Kumar, S., Mitsios, N., Slevin, M. and Kumar, P. (2007). Investigation of downstream target genes of PAX3c, PAX3e and PAX3g isoforms in melanocytes by microarray analysis. *Int J Cancer*, 120, 1223-31. [\[PubMed\]](#)

Wang, Q., Kumar, S., Slevin, M. and Kumar, P. (2006). Functional analysis of alternative isoforms of the transcription factor PAX3 in melanocytes in vitro. *Cancer Res*, 66, 8574-80. [\[PubMed\]](#)

Watanabe, A., Takeda, K., Ploplis, B. and Tachibana, M. (1998). Epistatic relationship between Waardenburg syndrome genes MITF and PAX3. *Nat Genet*, 18, 283-6. [\[PubMed\]](#)

Wiggan, O., Fadel, M. P. and Hamel, P. A. (2002). Pax3 induces cell aggregation and regulates phenotypic mesenchymal-epithelial interconversion. *J Cell Sci*, 115, 517-29. [\[PubMed\]](#)

Wiggan, O. and Hamel, P. A. (2002). Pax3 regulates morphogenetic cell behavior in vitro coincident with activation of a PCP/non-canonical Wnt-signaling cascade. *J Cell Sci*, 115, 531-41. [\[PubMed\]](#)

Wiggan, O., Taniguchi-Sidle, A. and Hamel, P. A. (1998). Interaction of the pRB-family proteins with factors containing paired-like homeodomains. *Oncogene*, 16, 227-36. [\[PubMed\]](#)

Wollnik, B., Tukel, T., Uyguner, O., Ghanbari, A., Kayserili, H., Emiroglu, M. and Yuksel-Apak, M. (2003). Homozygous and heterozygous inheritance of PAX3 mutations causes different types of Waardenburg

syndrome. Am J Med Genet A, 122A, 42-5. [\[PubMed\]](#)

Wu, M., Li, J., Engleka, K. A., Zhou, B., Lu, M. M., Plotkin, J. B. and Epstein, J. A. (2008). Persistent expression of Pax3 in the neural crest causes cleft palate and defective osteogenesis in mice. J Clin Invest, 118, 2076-87. [\[PubMed\]](#)

Yajima, I., Endo, K., Sato, S., Toyoda, R., Wada, H., Shibahara, S., Numakunai, T., Ikeo, K., Gojobori, T., Goding, C. R., et al. (2003). Cloning and functional analysis of ascidian Mitf in vivo: insights into the origin of vertebrate pigment cells. Mech Dev, 120, 1489-504. [\[PubMed\]](#)

Yang, G., Li, Y., Nishimura, E. K., Xin, H., Zhou, A., Guo, Y., Dong, L., Denning, M. F., Nickoloff, B. J. and Cui, R. (2008). Inhibition of PAX3 by TGF-beta modulates melanocyte viability. Mol Cell, 32, 554-63. [\[PubMed\]](#)

Zhou, H. M., Wang, J., Rogers, R. and Conway, S. J. (2008). Lineage-specific responses to reduced embryonic Pax3 expression levels. Dev Biol, 315, 369-82. [\[PubMed\]](#)

Zhu, B. K. and Pruitt, S. C. (2005). Determination of transcription factors and their possible roles in the regulation of Pax3 gene expression in the mouse B16 F1 melanoma cell line. Melanoma Res, 15, 363-73. [\[PubMed\]](#)

Zlotogora, J., Lerer, I., Bar-David, S., Ergaz, Z. and Abeliovich, D. (1995). Homozygosity for Waardenburg syndrome. Am J Hum Genet, 56, 1173-8. [\[PubMed\]](#)